NBDPS Abstractor's Instructions

Explanation/Glossary

BIRTH DEFECT & DEFINITION	The specific birth defect and its definition are listed. Other names for the defect are given as well as the defect group if applicable.
TYPES & DEFINITIONS	The types of the specific birth defect (if they exist) and their definitions are listed.
ICD-9-CM CODES	The ICD-9-CM codes for the specific birth defect are listed. This code is used for the discharge diagnoses and is generally less specific than a surveillance system's BPA or modified code.
INCLUSIONS	The standard inclusion criteria for the defect are listed. Any exceptions to the standard inclusions are listed in the table for that specific defect(s). The standard inclusions are as follows: 1) In general, case infants must have one or more of the specific birth defects listed in the tables. 2) The etiology of the defect(s) must be unknown or uncertain. 3) Cases can be live born or stillborn infants >= 20 weeks' gestation or 500 grams, or prenatally diagnosed and terminated fetuses at any gestational age. 4) Prenatally diagnosed and terminated fetuses that do not have a postnatal examination to confirm the defect will only be accepted as cases for a small number of defects. This information is listed in each individual table under the inclusion/exclusion boxes. While it is the goal of every Center to include terminations in the study, it may have to be phased in over time in certain Centers due to IRB issues. 5) Cardiac defects must be confirmed by echocardiography (echo), catheterization (cath), surgery or autopsy. Include cases diagnosed by prenatal ultrasound/echo only if done by a pediatric cardiologist or in a prenatal diagnosis center with expertise in
EXCLUSIONS	The standard exclusion criteria for the defect are listed. Any exceptions to the standard exclusions are listed in the table for that specific defect(s). The standard exclusions are as follows: 1) In general, fetuses/infants with chromosomal/microdeletion disorders or single gene disorders are excluded from the study. 2) Cardiac defects that are clinically-diagnosed, i.e., using only physical exam, chest radiography, ECG are excluded. 3) Autopsy cases in which maceration or disruption (from termination techniques) preclude definitive diagnosis are excluded. This does not apply to defects for which prenatal diagnosis is accepted under certain circumstances (see 4 and 5 under Inclusions).
ADDITIONAL BIRTH DEFECTS	Other birth defects that are often associated with the specific birth defect are listed; however, all birth defects should be listed for every case.
INFORMATION SOURCES-MED. RECORDS	The information sources in the medical records where the diagnosis/description of the specific birth defect would likely be found are listed. For some of the birth defects, a gold standard(s) is listed. If there is a discrepancy about the birth defect from the various sources, use the information from the gold standard(s).
DEFECT-SPECIFIC INFORMATION TO ABSTRACT	Other pertinent information to abstract for each specific birth defect is as follows: --chromosome results --syndrome diagnoses --family history of a birth defect (same or different) --maternal illnesses --maternal exposures to medications, etc. This information should be looked for and recorded (if present in the medical records) on all cases. If there is a particular reason to look more carefully for any of the above information, it will be listed.
ADD. COMMENTS	Helpful comments about each defect are listed.

Amnion rupture sequence

BIRTH DEFECT & DEFINITION	AMNION RUPTURE SEQUENCE--structural defects which are the consequence of tears or rupture in the amnion OTHER NAMES: amniotic bands; amniotic band sequence, syndrome, or disruption complex; Streeter bands
TYPES & DEFINITIONS	CONSTRICTION RINGS--soft tissue depressions or grooves encircling part of the body, usually a limb; pseudosyndactyly occuring when digits are compressed together AMPUTATIONS--the loss of a distal limb structure with the proximal structure intact CRANIOFACIAL ANOMALIES--oral clefts (usually not following normal closure planes), encephaloceles, anencephaly and various other disruptive defects of the cranium NOTE: Recently some authors have stated that craniofacial anomalies are not part of the spectrum of amniotic bands but are due to another disruptive mechanism. For this study, these defects will be included.
ICD-9-CM CODES	658.8
INCLUSIONS	standard
EXCLUSIONS	standard
ADDITIONAL BIRTH DEFECTS	pseudosyndactyly, limb hypoplasia, lymphedema or deformation such as clubfoot distal to constriction, oligohydramnios sequence (lung hypoplasia, flat facies, small chin, large ears, joint contractures), occasional attachment of the placenta or amnion to the body, short umbilical cord
INFORMATION SOURCES-MED. RECORDS	prenatal ultrasound, clinicians' (geneticist, orthopedist) and nurses' notes, placental examination GOLD STANDARD(S): clinicians' description
DEFECT-SPECIFIC INFORMATION TO ABSTRACT	1) description of the placenta 2) x-rays of affected limbs
ADD. COMMENTS	Occasionally distal limbs are very disrupted with amputations and reanastomoses of digits or other limb parts. It is difficult to make the diagnosis of amniotic bands in some instances (e.g., limb amputations above the foot without other signs of band anomalies) unless there has been a prenatal diagnosis of amniotic bands or other band-associated lesions are seen. Amputated digits do not have nails on the residual part of the digit. Constricted digits may be small or hypoplastic with nails but should have a noticeable

Anencephaly, craniorachischisis

BIRTH DEFECT & DEFINITION	ANENCEPHALY--partial or complete absence of the brain and skull OTHER NAMES: anencephalus, acrania, absent brain CRANIORACHISCHISIS--anencephaly with contiguous rachischisis (spine defect without meninges covering the neural tissue). Can be as limited as the cervical region or as extensive as the entire spine. DEFECT GROUP: neural tube defects (NTDs)
TYPES & DEFINITIONS	HOLOANENCEPHALY--defect extends thorough the foramen magnum (affects entire skull) MEROANENCEPHALY--defect limited to the anterior part of the brain and skull CRANIORACHISCHISIS WITH SPINAL RETROFLEXION--defect associated with severe anterior flexion of the spine NOTE: distinction between holo- and meroanencephaly rarely made in the medical record
ICD-9-CM CODES	ANENCEPHALY--740.0; CRANIORACHISCHISIS--740.1
INCLUSIONS	all cases including those cases prenatally diagnosed that do not have a postnatal examination to confirm the defect
EXCLUSIONS	standard
ADDITIONAL BIRTH DEFECTS	hypoplastic adrenal glands, small phallus and undescended testes, hydronephrosis, renal agenesis, clubfoot, developmental dysplasia of the hip, other limb deformations, corneal opacities, minor ear deformations NOTE: other birth defects such as orofacial clefts and heart defects are common in infants with anencephaly and craniorachischisis
INFORMATION SOURCES-MED. RECORDS	prenatal ultrasound, clinicians' or nurses' notes, autopsy GOLD STANDARD(S): no test is diagnostic but postnatal examination of the fetus/infant is very important to verify the diagnosis NOTE: maternal serum and amniotic fluid alpha-fetoprotein and acetylcholinesterase will be elevated since neural tissue is in contact with the amniotic fluid
DEFECT-SPECIFIC INFORMATION TO ABSTRACT	1) evidence of amniotic band sequence 2) note the most inferior aspect of bony spine defect for craniorachischisis
ADD. COMMENTS	AN: negative; ENCEPHALOS: brain; CRANIUM: skull; RHACHIS: spine; SCHISIS: fissure acrania is more properly used to describe absence of the skull without an underlying NTD

Anomalous pulmonary venous return

BIRTH DEFECT & DEFINITION	ANOMALOUS PULMONARY VENOUS RETURN--a condition in which a pulmonary vein or combination of pulmonary veins drains anomalously into the venous circulation of the right heart or the body instead of into the left heart. OTHER NAMES: anomalous pulmonary venous connection or drainage
TYPES & DEFINITIONS	TOTAL ANOMALOUS PULMONARY VENOUS RETURN/CONNECTION/DRAINAGE (TAPVR/TAPVC/TAPVD)--Failure of the 4 pulmonary veins to connect to the left atrium. Return of pulmonary blood occurs indirectly via other vessels, e.g. superior or inferior vena cava, right atrium. NOTE: Pulmonary blood returns to the heart via supra-diaphragmatic or infra-diaphragmatic routes. These details are not needed for coding purposes. PARTIAL ANOMALOUS PULMONARY VENOUS RETURN/CONNECTION/DRAINAGE (PAPVR/PAPVC/PAPVD)--Failure of 1-3 of the 4 pulmonary veins to connect to the left atrium. NOTE: As with TAPVR, location of the alternate pulmonary blood drainage is not relevant.
ICD-9-CM CODES	TAPVR: 747.41, PAPVR: 747.42
INCLUSIONS	standard
EXCLUSIONS	cases prenatally diagnosed that do not have a postnatal examination to confirm the diagnosis
ADDITIONAL BIRTH DEFECTS	Just about any defect can be found with this CHD.
INFORMATION SOURCES-MED. RECORDS	clinicians' or nurses' notes, pediatric cardiology consultation, pathology reports, cardiology diagnostic reports. GOLD STANDARD(S): echo (less reliable is prenatal ultrasound/echo), cath, surgery, autopsy
DEFECT-SPECIFIC INFORMATION TO ABSTRACT	standard
ADD. COMMENTS	TAPVR/PAPVR is called Class V in Clark's classification and is attributed to abnormal targeted growth, i.e., pulmonary veins fail to connect to the left atrium.

Anophthalmia, microphthalmia

BIRTH DEFECT & DEFINITION	ANOPHTHALMIA--total absence of the eye tissue or apparent absence of the globe in an orbit that otherwise contains normal adnexal structures. MICROPHTHALMIA--reduction in the volume of the eye, usually characterized by corneal diameter less than 10 mm or anteroposterior globe diameter less than 20 mm.
TYPES & DEFINITIONS	TRUE OR PRIMARY ANOPHTHALMIA--as above; occurs when there is complete failure of formation of the primary optic vesicle -usually bilateral -when unilateral, may have contralateral microphthalmia -verified only when histologic/microscopic exam shows that all ocular tissue is absent MICROPHTHALMIA--categories: colobomatous (uveal, iris, choroid and/or optic nerve) or noncolobomatous OTHER NAMES: nanophthalmia = microphthalmic eye with normal intraocular structures and is a distinct genetic malformation
ICD-9-CM CODES	ANOPHTHALMIA--743.00; MICROPHTHALMIA--743.10-743.12
INCLUSIONS	standard
EXCLUSIONS	--"small eyes" or "small palpebral fissures" unless there is confirmation of an- or microphthalmia --isolated microcornea with normal ocular size
ADDITIONAL BIRTH DEFECTS	other ocular malformations including, absence of the optic nerves and chiasm, rudimentary optic tracts, absent lacrimal puncta, ankyloblepharon, microphthalmia of opposite eye, congenital cyst of the orbit, congenital retinal detachments, secondary glaucoma, cataracts NOTE: Malformations of the brain (e.g., holoprosencephaly) and face often accompany anopthalmos and microphthalmos
INFORMATION SOURCES-MED. RECORDS	clinicians' exam with inspection; ophthalmology exam of the external eye structures and retinal exam; histopathology reports--serial sectioning of the orbit; ultrasonography or CT scan--precise measurement of the A-P axis
DEFECT-SPECIFIC INFORMATION TO ABSTRACT	ophthalmologic examination of other family members including parents of the proband for microphthalmia/ blindness
ADD. COMMENTS	MICRO: small; an: absent; OPHTHALMOS: eye anophthalmia and microphthalmia are frequent components of genetic syndromes NOTE: these conditions may be seen with the ending "ia", "os" or "us"

Anotia, microtia

BIRTH DEFECT & DEFINITION	ANOTIA--total absence of the external ear and canal MICROTIA--malformation or hypoplasia of the auricle, ranging from measurably small external ear with minimal structural abnormality, to an ear with major structural alteration with absent or blind-ending canal
TYPES & DEFINITIONS	Microtia (4 types: I - IV): Classification system of Meurman (modified from Marks) TYPE I--generally small ear that retains most of the overall structure of the normal auricle--similar to lop/ cup defect, auditory meatus is usually patent and defects of the ossicular chain are infrequent TYPE II--moderately severe anomaly with longitudinal mass of cartilage with some resemblance to pinna (rudimentary auricle will be hook-shaped, have an S-shape or question mark appearance) TYPE III--ear is a rudiment of soft tissue and the auricle has no resemblance to the normal pinna NOTE: Types I - III will occasionally be accompanied by a preauricular tag(s) . TYPE IV--complete absence of all external ear structures, anotia
ICD-9-CM CODES	ANOTIA--744.01; MICROTIA--744.23 NOTE: absence of the ear, congenital is included in the "other" code--744.09
INCLUSIONS	standard
EXCLUSIONS	--small ears that retain most of the normal structure or type I microtia --isolated atresia or stenosis of the external auditory canal
ADDITIONAL BIRTH DEFECTS	--associated middle ear anomalies or atresia with conductive hearing loss; sensorineural hearing loss --malposition, anomaly, hypoplasia of the facial nerve (with or without facial nerve palsy) --hypoplasia of ipsilateral mandible and face (secondary to malformation of the 1st branchial arch complex), narrowing of the osseous portion of eustachian tube, absence of torus tubarious, abnormal widening of the eustachian tube, inner ear abnormalities, absent or hypoplastic parotid gland, absence of ipsilateral tonsil --preauricular pits and appendage, branchial cysts, cleft palate --oculo-auriculo-vertebral (OAV) spectrum which includes Goldenhar syndrome and hemifacial microsomia has additional features such as mandibular hypoplasia, vertebral anomalies, macrostomia (lateral mouth cleft), epibulbar dermoids, variable heart and CNS defects NOTE WITH TYPES II AND III: ~100% associated external auditory canal atresia, 75% mandibular hypoplasia, 10% ipsilateral facial nerve weakness, and 14% hemifacial microsomia
INFORMATION SOURCES-MED. RECORDS	1) physical exam: clinicians' (otolaryngologist) or nurses' notes. 2) hearing evaluation (in infancy: brainstem evoked response and behavioral testing and impedance audiometry) 3) high resolution CT scan of the middle and inner ear (bilateral), if done
DEFECT-SPECIFIC INFORMATION TO ABSTRACT	1) laterality 2) hearing evaluation of parents and/or siblings 2) if done, evaluation of the external canal, middle and inner ear.
ADD. COMMENTS	AN: absent; MICRO: small; OTIA: ear anotia/microtia are frequent components of genetic syndromes (20-40%)

Atrioventricular septal defects

BIRTH DEFECT & DEFINITION	ATRIOVENTRICULAR SEPTAL DEFECTS--A variety of defects of the atrioventricular septa and the adjacent parts of the mitral and tricuspid valves. In the extreme virtually the entire atrial and ventricular septa may be missing. OTHER NAMES: AV canal, endocardial cushion defect, common atrioventricular orifices
TYPES & DEFINITIONS	ATRIAL SEPTAL DEFECT, PRIMUM TYPE (ASD1)--Hole (defect) in the lower portion of the atrial septum, towards the attachment of the AV valves. NOTE: Defect is usually accompanied by a congenital cleft of the mitral valve. Sometimes called "incomplete" AVC/ECD. "Common atrium" is like a high, large ASD primum so include in this category. INFLOW-TYPE (SUBTRICUSPID, CANAL-TYPE) VENTRICULAR SEPTAL DEFECT (VSDAVC)--Hole (defect) in the inflow (canal, subtricuspid) portion of the ventricular septum. This is an uncommon type of VSD. COMPLETE ATRIOVENTRICULAR CANAL (CAVC), ENDOCARDIAL CUSHION DEFECT (ECD), ATRIOVENTRICULAR SEPTAL DEFECT (AVSD)--Large hole (defect) involving both the inferior atrial septum and the superior ventricular septum resulting in a large canal/cushion defect. NOTE: The ventricular or atrial component of CAVC may be noted in cardiology reports. Don't record separately as VSD and ASD. Instead of a distinct tricuspid and mitral valve, there is a common atrioventricular valve. List but don't code valve anomaly separately.
ICD-9-CM CODES	ASD1--745.61, common atrium--745.69; VSDAVC--745.69; CAVC--745.60, 745.69
INCLUSIONS	standard
EXCLUSIONS	standard
ADDITIONAL BIRTH DEFECTS	Just about any defect can be found with this CHD.
INFORMATION SOURCES-MED. RECORDS	clinicians' or nurses' notes, pediatric cardiology consultation, pathology reports, cardiology diagnostic reports. GOLD STANDARD(S): echo (less reliable is prenatal ultrasound/echo), cath, surgery, autopsy
DEFECT-SPECIFIC INFORMATION TO ABSTRACT	standard
ADD. COMMENTS	These defects most likely are abnormalities of the structures derived from the embryonic endocardial cushions. They are grouped in Class IV of Clark's classification (attributed to abnormal extracellular matrix organization of the endocardial cushions). These defects have a well-known association with Down syndrome. Of DS individuals about 40% have congenital heart defects and about half of these have some type of AVC/ECD. Conversely, of patients with AVC/ECD, about 70% have DS.

Biliary atresia

BIRTH DEFECT & DEFINITION	BILIARY ATRESIA (EXTRAHEPATIC)--congenital absence of lumen of extrahepatic bile ducts
TYPES & DEFINITIONS	EXTRAHEPATIC--involving bile ducts which lie outside the liver
ICD-9-CM CODES	751.61
INCLUSIONS	extrahepatic--diagnosed by ultrasound, CT, MRI or HIDA scan; must have postnatal confirmation
EXCLUSIONS	intrahepatic biliary atresia (paucity of interlobular bile ducts) only documented postnatal acquisition of extrahepatic biliary atresia (i.e., post neonatal hepatitis)
ADDITIONAL BIRTH DEFECTS	other abnormalities of GI tract including absent gallbladder, patent urachus, heterotaxy
INFORMATION SOURCES-MED. RECORDS	clinicians' notes--particularly gastroenterology (GI) consultant, nurses' notes, liver function tests and elevated conjugated or direct bilirubin, ultrasound of abdomen, CT or MRI of abdomen, biliary excretion study (HIDA scan), surgical and pathology reports, autopsy GOLD STANDARD(S): surgical visualization (Kasai procedure) or pathology specimen
DEFECT-SPECIFIC INFORMATION TO ABSTRACT	viral titers
ADD. COMMENTS	--Alagille syndrome is a familial syndrome which includes intrahepatic biliary atresia, pulmonary artery stenosis and dysmorphic facial features. --Patients with biliary atresia may be said to have direct hyperbilirubinemia or jaundice. Phototherapy is not used to treat direct hyperbilirubinemia. Infants with extrahepatic biliary atresia usually have some involvement of the intrahepatic ducts also.

Cataracts, Glaucoma

BIRTH DEFECT & DEFINITION	CATARACTS/GLAUCOMA - Defects of the lens and anterior segment anomalies of the lens and anterior segment of the eye
TYPES & DEFINITIONS	CATARACT - A loss of transparency of the crystalline lens of the eye or of its capsule. Includes opacities affecting any part of the lens, including anterior, posterior and zonular cataracts. APHAKIA - Absence of the lens. LENS COLOBOMA - Absence of a portion of the lens. MICROSPHEROPHAKIA- A small, spherically-shaped lens. Can be seen as microphakia (small lens) or spherophakia (spherically-shaped lens). CONGENITAL GLAUCOMA OR BUPHTHALMOS- Disease of the eye characterized by increased intraocular pressure, often due to restricted outflow of the aqueous humor through Schlemm's canal. ANIRIDIA- Absence of the iris. ANTERIOR SEGMENT DYSGENESIS-A spectrum of eye malformations characterized by abnormal development of the anterior chamber angle and the iris. Types include Peters anomaly, Axenfeld anomaly and Rieger anomaly. Other names: anterior cleavage syndrome, mesodermal dysgenesis of the cornea and iris, primary dysgenesis mesodermalis of the iris, mesoectodermal dysgenesis of anterior segment, mesoectodermal dysgenesis of iris and cornea PETERS ANOMALY- Variable amounts of adhesions of the iris and lens to the central aspect of the posterior cornea. AXENFELD ANOMALY - Posterior embryotoxon (unusual prominence and forward displacement of the Schwalbe line) attached to dense bands of iris tissue. RIEGER ANOMALY (IRIDOGONIODYSGENESIS) - Iris stromal atrophy in addition to the Axenfeld anomaly.
ICD-9-CM CODES	congenital cataract, unspecified--743.30; capsular and subcapsular cataract--743.31; cortical and zonular cataract--743.32; nuclear cataract--743.33; total and subtotal cataract, congenital--743.34; congenital aphakia--743.35; anomalies of lens shape--743.36; buphthalmos, unspecified--743.20; simple buphthalmos--743.21; buphthalmos associated with other ocular anomalies; 743.44 - specified anomalies of anterior chamber, chamber angle, and related structures--743.44; aniridia--743.45; other specified anomalies of iris and ciliary body--743.46
INCLUSIONS	--These defects will be included in the NBDPS for births occurring after 1/1/2000 --All cases must include diagnosis by ophthalmologist within the first 12 months of life. --Isolated cases in which the family history appears consistent with a Mendelian inheritance pattern should be included, but notation regarding a possible single gene etiology should be made in the Clinical Reviewer's Comments.
EXCLUSIONS	--Cases that occur as a result of ophthalmologic surgery or trauma Note: With surgical aphakia diagnosis, search record for previous cataract diagnosis . -- Minor lens opacities (not clinically significant -- requiring no intervention or follow-up). -- Cases not examined by an ophthalmologist. -- Corneal clouding without other findings. -- Infants with Rieger syndrome, Peters Plus syndrome, Alagille syndrome, and Velocardiofacial/DiGeorge syndrome -- see additional comments below
ADDITIONAL BIRTH DEFECTS	Many birth defects can be associated with anterior segment and lens defects, particularly other anomalies of the head and central nervous system. Polycoria and ectopic pupil are frequently associated with these defects; medical record should be searched for possible eligible defects.
INFORMATION SOURCES-MED. RECORDS	Clinician's notes, ophthalmologic exams including both direct and indirect ophthalmoscopy, autopsy and pathology reports. Gold standard: pathologic/autopsy reports or exam by a pediatric ophthalmologist
DEFECT-SPECIFIC INFORMATION TO ABSTRACT	1) tests for congenital viral infection 2) chromosome anomalies 3) DNA mutations in genes known to cause cataracts when mutated (e.g. crystallins, PITX2, Lowe syndrome) 4) family history of eye defects
ADD. COMMENTS	--Rieger syndrome (OMIM 180500, 601499) is a autosomal dominant disorder that may include a variety of ophthalmologic conditions associated with glaucoma, including polycoria and Axenfeld anomaly. Peters-Plus or Peters anomaly with short-limbed dwarfism (OMIM 261540) is an autosomal recessive disorder characterized by Peters anomaly plus short limbs, cleft lip +/- palate, and mental retardation. Posterior embryotoxon and other types of anterior segment dysgenesis are frequent eye findings in Alagille syndrome (OMIM 118450) and posterior embryotoxon is also often found in the Velocardiofacial/DiGeorge syndrome (OMIM 192430; 188400).

Choanal atresia

BIRTH DEFECT & DEFINITION	CHOANAL ATRESIA--congenital obstruction of the posterior choana or choanae. Choana=the opening into the nasopharynx of the nasal cavity on either side.
TYPES & DEFINITIONS	unilateral or bilateral membranous membranous with a bony rim completely bony
ICD-9-CM CODES	choanal atresia--748.0
INCLUSIONS	all types; case must have at least unilateral choanal atresia
EXCLUSIONS	choanal stenosis only
ADDITIONAL BIRTH DEFECTS	CHARGE association= Coloboma, Heart defect, Atresia choanae, Retarded growth and development, Genital hypoplasia, Ear anomalies or deafness
INFORMATION SOURCES-MED. RECORDS	clinicians' and nurses' notes, ENT consult, CT scan, operative/path reports, autopsy GOLD STANDARD(S): operative/path reports, autopsy
DEFECT-SPECIFIC INFORMATION TO ABSTRACT	standard
ADD. COMMENTS	Respiratory distress may be noted in delivery room (if bilateral atresia) and feeding tube will not pass through nares. Most choanal atresia is posterior choanal atresia--note if the defect is described as anterior.

Cleft lip +/- palate

BIRTH DEFECT & DEFINITION	CLEFT LIP +/- PALATE--incomplete closure of the lip; often accompanied by a maxillary alveolar (gum) defect and/or cleft palate ; maxillary alveolar defect may be a complete cleft that is continuous with the cleft palate, or it may be limited to a notch on the gum; cleft lip may be unilateral, bilateral, or median (distinguished from bilateral cleft lip by agenesis of premaxilla)
TYPES & DEFINITIONS	COMPLETE CLEFT LIP--defect extends through the entirety of the lip and the nasal floor; may be unilateral or bilateral; usually associated with a more severe nasal deformation INCOMPLETE CLEFT LIP--defect of lip that does not extend into the nasal floor; may be unilateral or bilateral; there may be an incomplete cleft lip on one side and a complete cleft lip on the other side PSEUDOCLEFT LIP--abnormal linear thickening or depressed groove of skin, or subtle scar-like pigmentary difference paralleling the philtral ridge on the affected side; may be associated with slight notch of the vermillion or a mild slouching of the alar cartilage
ICD-9-CM CODES	CLEFT LIP--749.10-14; CLEFT LIP WITH PALATE--749.20-25
INCLUSIONS	standard
EXCLUSIONS	--If cleft palate is associated with any type of cleft lip, it is classified as a cleft lip, not cleft palate. --pseudocleft lip; Tessier type facial clefts; oblique facial clefts
ADDITIONAL BIRTH DEFECTS	standard
INFORMATION SOURCES-MED. RECORDS	prenatal ultrasonography; clinicians' or nurses' notes, autopsy, craniofacial centers, surgical notes GOLD STANDARD(S): notes from craniofacial centers, surgical or autopsy notes
DEFECT-SPECIFIC INFORMATION TO ABSTRACT	1) describe status of lip and palate separately 2) describe complete vs. incomplete status for right and left sides 3) describe laterality (right, left, median); if laterality not stated, code as NOS 4) evidence of amniotic bands
ADD. COMMENTS	Note: If complete or incomplete unilateral cleft lip is accompanied by a pseudocleft lip on the contralateral side, code as a bilateral cleft lip.

Cleft palate

BIRTH DEFECT & DEFINITION	CLEFT PALATE--hole in roof of the mouth; incomplete fusion of the palatal shelves; may be limited to soft palate or also extend onto hard palate. If cleft palate is associated with cleft lip, it is classified as a cleft lip.
TYPES & DEFINITIONS	SUBMUCOUS CLEFT PALATE--defect of the soft palate with mucosa or a reduced, thin muscle layer bridging the midline; difficult to diagnose clinically in 1st year; often associated with a bifid uvula PIERRE ROBIN ANOMALY (SEQUENCE)--combination of micrognathia, cleft palate, glossoptosis (tongue falls back into pharynx)
ICD-9-CM CODES	749.0
INCLUSIONS	standard
EXCLUSIONS	--If cleft palate is associated with any type of cleft lip, it is classified as a cleft lip, NOT cleft palate. --bifid uvula without overt cleft palate; submucous cleft palate
ADDITIONAL BIRTH DEFECTS	Pierre Robin anomaly
INFORMATION SOURCES-MED. RECORDS	clinicians' or nurses' notes, autopsy, craniofacial centers, surgical notes GOLD STANDARD(S): notes from craniofacial centers, surgery or autopsy
DEFECT-SPECIFIC INFORMATION TO ABSTRACT	1) note if lip is stated to be normal 2) Pierre Robin anomaly diagnosed? 3) verbatim description of defect, including unilateral, bilateral, or central (if stated)
ADD. COMMENTS	Designation of U-shaped vs. V-shaped cleft is not helpful.

Conotruncal heart defects

BIRTH DEFECT & DEFINITION	CONOTRUNCAL HEART DEFECTS--anomalies of the outflow tract of the heart OTHER NAMES: outflow tract anomalies
TYPES & DEFINITIONS	MALALIGNMENT-TYPE (TYPE 1, SUBARTERIAL, CONOVENTRICULAR) VENTRICULAR SEPTAL DEFECT (VSDMAL)--Hole (defect) high in the ventricular septum, under the aorta. NOTE: Only defect in the conotruncal category which does not involve the aortic arch. DOUBLE-OUTLET RIGHT VENTRICLE (DORV)--Both the pulmonary artery and the aorta arise from right ventricle, usually accompanied by a ventricular septal defect (VSD). This defect is usually associated with other CHD's (e.g., VSD, pulmonic stenosis, COA, complex single ventricle). NOTE: The great arteries can be normally related (when the VSD is subaortic) or transposed (when the VSD is subpulmonary), so specify. DORV is not dTGA. Special sub-type is Taussig-Bing anomaly (subPS, COA). TETRALOGY OF FALLOT (TOF, TET)--Tetralogy = malalignment-type VSD creates subvalvar pulmonic stenosis, overriding of the aorta, and right ventricular hypertrophy. NOTE: Do not code VSD and pulmonic stenosis separately. "Pentalogy of Fallot" (TOF + ASD2) is an archaic term. PULMONARY ATRESIA WITH VSD (PA/VSD, TETRALOGY WITH PULMONARY ATRESIA)--Absent connection from the right ventricle to the pulmonary artery and the aorta, usually with malignment-type VSD. NOTE: Distinguish from PA with intact ventricular septum (see obstructive heart defects). When VSD accompanies PA, this complex usually belongs in the tetralogy family (TOF/PA). Alternative archaic terms are Truncus, type 4 or pseudotruncus. If PA/VSD are both present and anatomic details of the VSD/aorta are not described in the echocardiogram report, e.g., "membranous/malalignment-type," or if the VSD is described as "muscular," the Pulmonary Atresia with VSD (not TOF variant 747.310) code should be used (see obstructive heart defects). TRUNCUS ARTERIOSUS (TA)--Single common arterial trunk instead of separate pulmonary artery and aorta, almost always associated with a malalignment-type VSD. NOTE: There are subtypes 1, 2, 3 based on the pattern of truncal branching. No need to specify type. DEXTRO-TRANSPOSITION OF GREAT ARTERIES (DTGA, DTGV)--Transposed great arteries such that the pulmonary artery arises from the left ventricle and the aorta arises from the right ventricle. May be isolated or with other CHDs (e.g., VSD, pulmonic stenosis). INTERRUPTED AORTIC ARCH, TYPE B (IAA, TYPE B)--Interruption of the descending aorta distal to the left carotid/proximal to the left subclavian artery, almost always associated with a malalignment-type VSD. NOTE: Interruption can occur at other locations along the arch, e.g. type A. Specify type.
ICD-9-CM CODES	VSDMAL--745.4; DORV--745.11; TOF, TET--745.2; PA/VSD, TOF WITH PA--747.3 and 745.2; TA--745.0; DTGA--745.10; IAA, TYPE B--747.11
INCLUSIONS	Include infants who are NEGATIVE or NOT TESTED for 22q11 deletion.
EXCLUSIONS	Exclude infants who are POSITIVE for 22q11 deletion.
ADDITIONAL BIRTH DEFECTS	Just about any defect can be found with these CHDs. Of all CHDs, dTGA is the exception--most likely to be an isolated CHD.
INFORMATION SOURCES-MED. RECORDS	clinicians' or nurses' notes, pediatric cardiology consultation, pathology reports, cardiology diagnostic reports. GOLD STANDARD(S): echo (less reliable is prenatal ultrasound/echo), cath, surgery, autopsy
DEFECT-SPECIFIC INFORMATION TO ABSTRACT	1) FISH analysis for 22q11 deletion (DiGeorge sequence, Velo-cardio-facial syndrome) 2) maternal exposure to drugs (e.g., retinoic acid) 3) maternal illness (e.g. maternal diabetes, fever, flu)
ADD. COMMENTS	This group is called Class I in Clark's classification (as well as aortic arch anomalies not included in this project). These defects are attributed to abnormal mesenchymal tissue migration (neural crest). Associated with 3rd & 4th branchial arch anomalies.

Craniosynostosis

BIRTH DEFECT & DEFINITION	CRANIOSYNOSTOSIS--premature closure of one or several cranial sutures (connective tissue membranes that separate the bones of the developing skull) OTHER NAME: craniostenosis
TYPES & DEFINITIONS	1) Cranial shapes that may or may not result from craniosynostosis: DOLICHOCEPHALY/SCAPHOCEPHALY--long, wedge-shaped skull with a prominent forehead and occiput resulting from premature closure of sagittal suture BRACHYCEPHALY--high, wide, short skull resulting from premature fusion of coronal sutures OXYCEPHALY/TURRICEPHALY/ACROCEPHALY--tall, tower-like skull (sometimes pointed) resulting from premature fusion of coronal and usually sagittal sutures PLAGIOCEPHALY--asymmetric skull shape which can result from unilateral closure of coronal and/or lambdoidal suture TRIGONOCEPHALY--triangular-shaped skull resulting from premature closure of metopic suture NOTE: all cases must have craniosynostosis for inclusion 2) Major sutures involved: sagittal, coronal, lambdoidal, metopic
ICD-9-CM CODES	756.0
INCLUSIONS	must have confirmation of craniosynostosis diagnosis by postnatal imaging (head CT is preferable; plain films at a minimum)
EXCLUSIONS	deformational plagiocephaly without synostosis and other abnormal head shapes described above without craniosynostosis
ADDITIONAL BIRTH DEFECTS	craniofacial anomalies
INFORMATION SOURCES-MED. RECORDS	prenatal ultrasound, postnatal X-ray and/or head CT, clinicians' and nurses' notes, plastic surgery and/or neurosurgery consults, operative/path reports, autopsy GOLD STANDARD(S): CT scan
DEFECT-SPECIFIC INFORMATION TO ABSTRACT	1) which suture(s) involved 2) complete or partial 3) laterality
ADD. COMMENTS	--Craniosynostosis is seen in many syndromes such as the acrocephalosyndactylies, in which there are limb abnormalities such as syndactyly. --A particularly severe form of craniosynostosis of multiple sutures is called cloverleaf skull or Kleeblattschädel. This condition is usually (85%) associated with a syndrome diagnosis.

Dandy-Walker malformation

BIRTH DEFECT & DEFINITION	DANDY-WALKER MALFORMATION--malformation of the cerebellum consisting of the triad: 1) hypoplasia or absence of the vermis; 2) upward displacement of the falx, lateral sinuses, and torcular; 3) a large thin-walled retrocerebellar cystic-like dilatation of the roof of the 4th ventricle. Some researchers consider isolated hypoplasia of the cerebellar vermis to be part of the Dandy-Walker spectrum. NOTE: Dandy-Walker variant is an inappropriate term which has been used for isolated atresia of the cerebellar foramina, for an enlarged cisterna magna and for cysts that do not communicate with the 4th ventricle.
TYPES & DEFINITIONS	N/A
ICD-9-CM CODES	Dandy-Walker malformation--742.3; cerebellar hypoplasia--742.2
INCLUSIONS	isolated cerebellar vermis hypoplasia
EXCLUSIONS	standard
ADDITIONAL BIRTH DEFECTS	--Hydrocephalus does not always occur in Dandy-Walker malformation, but is a frequent secondary complication --Common presenting sign is a macrocrania (large head) often with prominent occiput; neurologic signs, including signs of intracranial pressure and hypotonia --Additional CNS abnormalities including agenesis of the inferior cerebellar vermis and corpus callosum, abnormal inferior olives, enlarged cisterna magna, ventriculomegaly - Associated extracranial malformations including multicystic kidney disease, congenital hepatic fibrosis, facial angioma, cardiovascular and digital anomalies
INFORMATION SOURCES-MED. RECORDS	physical exam (including transillumination of the posterior fossae); CT scan, radionucleotide scanning and sonography, MRI scan, prenatal ultrasound, surgical report, autopsy GOLD STANDARD(S): MRI
DEFECT-SPECIFIC INFORMATION TO ABSTRACT	verbatim description of brain imaging studies
ADD. COMMENTS	Collect case even if the term Dandy-Walker variant is used and submit to clinical reviewer for decision on case status.

Diaphragmatic hernia

BIRTH DEFECT & DEFINITION	DIAPHRAGMATIC DEFECTS (HERNIA)--incomplete formation of the diaphragm in which some portion of the abdominal contents herniates into the thoracic cavity. OTHER NAMES: absence of diaphragm, hemidiaphragm
TYPES & DEFINITIONS	Posterolateral hernia = Bochdalek hernia Anterior hernia = Morgagni hernia Paraesophageal
ICD-9-CM CODES	756.6
INCLUSIONS	all cases including those prenatally diagnosed (only if bowel was documented in the chest by prenatal ultrasound)
EXCLUSIONS	eventration of the diaphragm--not a true herniation but an upward displacement of abdominal contents into an out pouching or sac-like structure of the diaphragm resulting from a weakness or absence of diaphragmatic musculature
ADDITIONAL BIRTH DEFECTS	pulmonary hypoplasia heart and mediastinal structures often shifted to the contralateral side (usually right) of the defect
INFORMATION SOURCES-MED. RECORDS	prenatal ultrasound, postnatal x-ray, contrast studies of the bowel (Morgagni hernia), clinicians' or nurses' notes, surgery consult, operative/path reports, autopsy, death certificate, fetal death certificate
DEFECT-SPECIFIC INFORMATION TO ABSTRACT	standard
ADD. COMMENTS	none

Ebstein malformation

BIRTH DEFECT & DEFINITION	EBSTEIN MALFORMATION OR ANOMALY--downward displacement of the tricuspid valve into the right vetricle ("atrialization" of ventricles). The tricuspid valve is usually hypoplastic and regurgitant.
TYPES & DEFINITIONS	Type is not usually specified; description preferred because of highly variable degrees of severity.
ICD-9-CM CODES	746.2
INCLUSIONS	standard
EXCLUSIONS	standard
ADDITIONAL BIRTH DEFECTS	Just about any defect can be found with this CHD.
INFORMATION SOURCES-MED. RECORDS	clinicians' or nurses' notes, pediatric cardiology consultation, pathology reports, cardiology diagnostic reports. GOLD STANDARD(S): echo (less reliable is prenatal ultrasound/echo), cath, surgery, autopsy
DEFECT-SPECIFIC INFORMATION TO ABSTRACT	maternal exposures--Ebstein anomaly has long been associated with lithium exposure; however, current thought is that the magnitude of the association is extremely small.
ADD. COMMENTS	Ebstein anomaly falls in Class III of Clark's classification (putatively attributed to abnormal cell death). There is little experimental evidence for this grouping.

Encephalocele, cranial meningocele,

BIRTH DEFECT & DEFINITION	ENCEPHALOCELE--a saccular herniation through the cranium, usually skin-covered, which contains brain tissue CRANIAL MENINGOCELE--a saccular herniation from the cranium, often skin-covered, which contains meninges and cerebrospinal fluid but no brain tissue ENCEPHALOMYELOCELE--a saccular herniation similar to an encephalocele that spans a portion of both the cranium and the spine OTHER NAMES: cephalocele (includes both encephalocele and cranial meningocele), meningo- encephalocele, ventriculocele, encephalocystomeningocele, hydranencephalocele DEFECT GROUP: neural tube defects (NTDs)
TYPES & DEFINITIONS	ENCEPHALOCELE, CRANIAL MENINGOCELE--can affect any part of the cranium externally (nasal, frontal, parietal, occipital or combinations) or internally (sphenoid, orbital, maxillary, ethmoid, pharyngeal, or combinations)
ICD-9-CM CODES	ENCEPHALOCELE--742.0
INCLUSIONS	all cases including those cases prenatally diagnosed that do not have a postnatal examination to confirm the defect
EXCLUSIONS	standard
ADDITIONAL BIRTH DEFECTS	Any number of birth defects can be seen with cephaloceles including other CNS anomalies such as absence of the corpus callosum and Dandy-Walker malformation.
INFORMATION SOURCES-MED. RECORDS	prenatal ultrasound, clinicians' or nurses' notes, surgery notes, autopsy, CT or MRI scans GOLD STANDARD(S): TYPE--surgical or autopsy report, MR or CT scan NOTE: maternal serum and amniotic fluid alpha-fetoprotein and acetylcholinesterase usually not be elevated since neural tissue is not in contact with the amniotic fluid
DEFECT-SPECIFIC INFORMATION TO ABSTRACT	amniotic band sequence
ADD. COMMENTS	ENCEPHALO: brain; MYELO: spinal cord; CELE: hernia Meckel-Gruber syndrome is an genetic disorder that has occipital encephalocele, postaxial polydactyly, and polycystic kidneys as main features.

Esophageal atresia +/- TE fistula

BIRTH DEFECT & DEFINITION	ESOPHAGEAL ATRESIA +/- TRACHEOESOPHAGEAL FISTULA (T-E FISTULA, TEF)--congenital complete discontinuity of the lumen of the esophagus resulting in a blind esophageal pouch occurring with or without an abnormal communication between the esophagus and trachea.
TYPES & DEFINITIONS	There are several classification schemas. In 90% of cases the upper esophagus ends in a blind pouch and the lower segment forms a fistula with the trachea.
ICD-9-CM CODES	750.3
INCLUSIONS	standard
EXCLUSIONS	--TE fistula without esophageal atresia --esophageal stenosis --trachea atresia -- tracheoesophageal cleft
ADDITIONAL BIRTH DEFECTS	VACTERL association (Vertebral anomalies, Anal atresia, Cardiac defects, TracheoEsophageal fistula, Renal anomalies, Limb anomalies)
INFORMATION SOURCES-MED. RECORDS	prenatal ultrasound (polyhydramnios, nonvisualization of the stomach), x-ray after placement of a radiopaque feeding tube or contrast material, surgical notes, autopsy GOLD STANDARD(S): surgery or autopsy
DEFECT-SPECIFIC INFORMATION TO ABSTRACT	standard
ADD. COMMENTS	Diagnosis of TE fistula without esophageal atresia may be delayed and thus is not included in the case definition.

Exstrophy, bladder

BIRTH DEFECT & DEFINITION	BLADDER EXSTROPHY--a defect in the lower abdominal wall in which the bladder mucosa (lining) is exposed OTHER NAMES: extroversion of bladder, epispadias-exstrophy complex, vesical exstrophy, ectopia vesicae
TYPES & DEFINITIONS	CLASSIC--urinary tract is open anteriorly from urethral meatus to umbilicus. There is wide separation of pubic symphysis, rectus abdominus muscles, and rectus fascia. There is eversion/exposure of the posterior bladder wall. The genitalia of both genders can be involved and may be bifid or duplicated. VARIANTS--rare and more common in females as compared to the classic form where males predominate. They include superior vesical fistula, closed exstrophy, duplicate exstrophy, pseudoexstrophy, inferior vesicle, epispadias (penopubic and balanic penile). NOTE: If variants are described, collect information for clinician decision re case inclusion with the exception of isolated epispadias which is excluded.
ICD-9-CM CODES	753.5
INCLUSIONS	standard
EXCLUSIONS	isolated epispadias
ADDITIONAL BIRTH DEFECTS	"ambiguous genitalia" may be charted if obvious scrotum and testes are not present; epispadias almost uniformly present; occasional defects of the sacrum or hindgut
INFORMATION SOURCES-MED. RECORDS	prenatal ultrasound; increased maternal serum or amniotic fluid alpha-fetoprotein (AFP); clinicians' and nurses' notes; abdominal X-ray, ultrasound, CT, MRI; sugical notes; various radiology studies using dyes injected into the presumed urethral orifice; surgical or pathology reports; autopsy GOLD STANDARD(S): surgical description of the lesion
DEFECT-SPECIFIC INFORMATION TO ABSTRACT	presence of a patent anus
ADD. COMMENTS	Chart may refer to fluid leaking from the site--this may be urine. In essence, the anterior wall of the bladder and lower abdomen is missing. 3x more common in males than in females. Testes (in males) are likely to be in their normal location. Anus is patent, as opposed to cloacal exstrophy, where anus is imperforate.

Exstrophy, cloacal

BIRTH DEFECT & DEFINITION	CLOACAL EXSTROPHY--congenital persistence of a common cloacal cavity into which gut, urethra, and reproductive tracts open, with exstrophy of the cavity. OTHER NAMES: exstrophia splanchnica, persistent cloaca
TYPES & DEFINITIONS	CLASSIC--most severe manifestation of the epispadias-exstrophy spectrum of ventral body wall defects. Exstrophy of the bladder with intestinal epithelium between the hemibladders, phallic separation with epispadias, separation of the pubic arch anteriorly, and rudimentary hindgut with imperforate anus VARIANTS--account for slightly less than ½ of all cases. Most common are pseudoexstrophy and closed exstrophy in which the abdominal wall is partially or completely formed and skin-covered.
ICD-9-CM CODES	751.5
INCLUSIONS	standard
EXCLUSIONS	standard
ADDITIONAL BIRTH DEFECTS	Omphalocele-Exstrophy-Imperforate anus-Spina bifida (OEIS complex); intestinal/colonic abnormalities; hip and lower limb anomalies; absent kidneys or gallbladder
INFORMATION SOURCES-MED. RECORDS	prenatal ultrasound; increased maternal serum or amniotic fluid alpha-fetoprotein; clinicians' and nurses' notes; abdominal x-ray, CT, ultrasound, MRI; surgical notes; various radiology studies using dyes injected into the presumed urethral orifice; autopsy. GOLD STANDARD(S): surgical description of the lesion or autopsy
DEFECT-SPECIFIC INFORMATION TO ABSTRACT	presence of amniotic band sequence or amniotic attachment to lesion
ADD. COMMENTS	May initially be diagnosed as exstrophy of the bladder, with modification of the diagnosis with surgery or detailed diagnostic studies. Imperforate anus is part of cloacal exstrophy and not part of bladder exstrophy. May be 2x as common in males.

Gastroschisis

BIRTH DEFECT & DEFINITION	GASTROSCHISIS--congenital fissure of the anterior abdominal wall, lateral to the umbilicus, usually to the right, with a small bridge of skin separating the defect from the umbilicus. Accompanied by herniation of the small, and part of the large, intestines, and occasionally other abdominal organs, into the amniotic cavity, and lacking a protective membrane
TYPES & DEFINITIONS	LIMB-BODY WALL COMPLEX: disruption complex involving lateral body wall defect, limb reduction defect, neural tube defects, heart and other anomalies
ICD-9-CM CODES	756.79
INCLUSIONS	all cases including those prenatally diagnosed if high resolution ultrasound was done and umbilicus was visualized
EXCLUSIONS	standard
ADDITIONAL BIRTH DEFECTS	limb-body wall complex small bowel atresia, "apple peel" anomaly of small bowel, microcolon porencephaly, other vascular disruptions
INFORMATION SOURCES-MED. RECORDS	prenatal ultrasound, clinicians' or nurses' notes, autopsy, surgery notes GOLD STANDARD(S): surgery notes or autopsy NOTE: maternal serum and/or amniotic fluid alpha-fetoprotein may be elevated
DEFECT-SPECIFIC INFORMATION TO ABSTRACT	1) size (diameter) and location of abdominal wall defect R or L of umbilicus) 2) # of umbilical vessels 3) which organs extruded through abdominal wall defect 4) other intestinal problems (e.g. bowel atresia) 5) note if amniotic band sequence diagnosed
ADD. COMMENTS	It is essential to differentiate gastroschisis from omphalocele, in which the protruding abdominal contents are herniated into the umbilical stalk and are covered by a membrane that is continuous with the abdominal skin. An omphalocele RARELY ruptures. In gastroschisis, the extruded abdominal contents may be matted and covered by a thick fibrous material, but this membrane does not

Heterotaxia

BIRTH DEFECT & DEFINITION	HETEROTAXIA--alteration of the normal laterality of organs in chest (e.g., heart, bronchi, lungs) and abdomen (e.g., spleen, liver). One form of heterotaxia (situs ambiguus) is particularly associated with complex heart anomalies. OTHER NAMES: laterality defects, isomerism, malposition
TYPES & DEFINITIONS	SITUS INVERSUS (SI)--Reversed laterality of organs in chest (heart, bronchi) and/or abdomen (spleen and stomach on right, liver on left) , i.e., left and right are essentially swapped in some internal organs. When both chest and abdominal organs are involved, the situation is termed SITUS INVERSUS (SI) TOTALIS. SITUS INVERSUS ABDOMINUS can occur without congenital heart defects but would not be included in the study. SITUS AMBIGUUS--The laterality is neither solitus (normal) nor inversus (reversed), but several organs are either bilaterally 'right' or 'left' . This situation is also called bilateral sideness. Two categories are recognized: --BILATERAL RIGHTEDNESS, OR RIGHT ISOMERISM--Bronchi and atrial appendages have bilateral right morphology. The liver is in the midline, and organs that are usually on the left may be absent [e.g., the spleen--ASPLENIA COMPLEX (see below), or some pulmonary veins]. --BILATERAL LEFTEDNESS OR LEFT ISOMERISM--Bronchi and atrial appendages have bilateral left morphology. The liver is in the midline, and organs that usually on the left may be present in multiple copies [e.g., the spleen--POLYSPLENIA COMPLEX (see below)], and some organs usually on the right may be absent (e.g., inferior vena cava).
ICD-9-CM CODES	759.3
INCLUSIONS	standard
EXCLUSIONS	--isolated dextrocardia --isolated situs inversus abdominus --situs inversus abdominis associated with cardiac defects (with the exception of PFO, PDA, muscular VSDs, and functional defects such as mild valvular regurgitation, cardiomegaly, etc.) should be INCLUDED in this category, even if the cardiac defect is not an NBDPS-eligible defect.
ADDITIONAL BIRTH DEFECTS	--gastrointestinal (in addition to liver and spleen), e.g.. biliary atresia, malrotation, intestinal atresia.
INFORMATION SOURCES-MED. RECORDS	clinicians' or nurses' notes, pediatric cardiology consultation, pathology reports, cardiology diagnostic reports. GOLD STANDARD(S): echo (less reliable is prenatal ultrasound/echo), cath, surgery, autopsy
DEFECT-SPECIFIC INFORMATION TO ABSTRACT	Heterotaxias are complex phenotypes. Appropriate coding and classification depend critically on the careful reporting of a spectrum of anomalies. When facing a possible case of heterotaxia, please review carefully any mention of the following structures: Great veins (mainly inferior vena cava and pulmonary veins, but also azygos and superior vena cava). Note in particular: --Location and relationship of abdominal IVC and aorta (e.g., "IVC and aorta to the left", or "on the same side of spine") --Interrupted inferior vena cava; azygous continuation to SVC --Anomalous pulmonary venous return, --Number of superior venae cavae (SVC), or presence of left SVC Atria --Note reference to left or right atrial isomerism --Note unusually large ASD (common atrium) Lung morphology --Note number of lobes of the right and left lungs, and the morphology of bronchi. Spleen --Note if no spleen, one spleen on right or left, or multiple spleens; note the examinations used to detect spleen (e.g., echo, scan, MRI, physical exam), and their findings. --Be specific: say ' MRI shows no spleen' rather than summarize as asplenia. Please note that some people (including doctors) use asplenia or polysplenia as summary diagnosis of isomerism (polysplenia, left; asplenia, right) regardless of spleen status. Heart rhythm --Note arrhythmias, particularly heart block; also note if heart rhythm is normal (heart block can be a sign of left isomerism) Look systematically for heart defects (particularly AV canal, DILV, TGA, DORV). Note whether a transposition is D or L. Note outflow tract obstructions. Also note position of heart in the chest (levocardia, dextrocardia). It may be tedious work to record these heart anomalies, but it pays off, as some defect combinations are suggestive of specific heterotaxias.
ADD. COMMENTS	Other terms in use (avoid if possible): IVEMARK SYNDROME--Used in conditions resembling bilateral right sidedness. ASPLENIA COMPLEX--Refers specifically to situs ambiguus with right isomerism (see above), when the spleen is absent. However, this is a confusing term because situs ambiguus with right morphology can coexist with a spleen. POLYSPLENIA COMPLEX--Refers specifically to situs ambiguus with left isomerism (see above), with "bilateral leftedness" of bronchi and atrial appendages, when in addition many small spleens are present.

Holoprosencephaly

BIRTH DEFECT & DEFINITION	HOLOPROSENCEPHALY--variable degrees of failure of horizontal and vertical cleavage of the prosencephalon (early single-ventricle forebrain) into the paired telencephalon and diencephalon OTHER NAMES: holotelencephaly, cyclopia, proboscis, ethmocephaly
TYPES & DEFINITIONS	ANATOMIC SUBGROUPS: ALOBAR--no cleavage of prosencephalon; no temporal or occipital horns of ventricles; absent 3rd ventricle, fused thalami; olfactory bulbs and corpus callosum always absent SEMILOBAR--partial cleavage of the prosencephalon; development of the interhemispheric fissure proceeds posterior to anterior, so it is not present anteriorly; temporal horns have developed, rudimentary 3rd ventricle, variably developed occipital horns and posterior fissure and falx; corpus callosum usually very hypoplastic; olfactory bulbs hypoplastic or absent LOBAR--difficult to detect; distinct interhemispheric fissure present and brain has well-formed lobes; lateral ventricles are closely apposed and narrow; corpus callosum may be absent, hypoplastic, or normal; olfactory bulbs may be absent or hypoplastic ASSOCIATED FACIAL PHENOTYPES: CYCLOPIA--single midline orbit that can be anophthalmic, monophthalmic, or contain synophthalmic ocular structures. Proboscis (blind-ended tubular structure) is usually present, usually above the orbit, but no nasal structures (arhinia) ETHMOCEPHALY--least common; proboscis separates 2 severely hypoteloric orbits, usually with marked microphthalmia and absent nasal structures (arhinia) CEBOCEPHALY--small single blind-ending nostril nose below hypoteloric, usually hypoplastic orbits premaxillary agenesis (median cleft lip)-- midline cleft lip with absent premaxilla; nose small and flat, and ocular hypotelorism common LESS SEVERE FACIAL FEATURES--hypotelorism or hypertelorism, flat nose, unilateral or bilateral cleft lip
ICD-9-CM CODES	HOLOPROSENCEPHALY--742.2; CYCLOPIA--759.89
INCLUSIONS	standard
EXCLUSIONS	aprosencephaly, atelencephaly, hydranencephaly, arhinencephaly without holoprosencephaly
ADDITIONAL BIRTH DEFECTS	microcephaly; facial phenotypes described above, plus trigonocephaly; arhinencephaly (agenesis of olfactory bulbs); hypotelorism; unilateral cleft lip; agnathia
INFORMATION SOURCES-MED. RECORDS	prenatal ultrasound of brain, clinicians' or nurses' notes, autopsy, brain CT or MRI report GOLD STANDARD(S): CT or MRI scan, autopsy
DEFECT-SPECIFIC INFORMATION TO ABSTRACT	1) holoprosencephaly has a continuum of anatomic severity, and when possible, should be subtyped into alobar, semi-lobar and lobar anatomic subtypes 2) associated facial features (cyclopia, cebocephaly, ethmocephaly, premaxillary agenesis, cleft lip) 3) associated brain malformations (hydrocephalus, agyria, microgyria, heterotopias) 4) head circumference or presence of microcephaly
ADD. COMMENTS	Arhinencephaly may occur with holoprosencephaly, but can also occur as an isolated defect.

Hydrocephalus

BIRTH DEFECT & DEFINITION	HYDROCEPHALUS--increase in intracranial pressure caused by an increase in the intracranial cerebrospinal fluid (CSF) space relative to the space occupied by the brain tissue, and usually refers to internal hydrocephalus in which all or part of the ventricular
TYPES & DEFINITIONS	AQUEDUCTAL STENOSIS--cerebral aqueduct (aqueduct of Sylvius) is narrowed and/or consisting of several minute channels with blockage of CSF circulation, which results in dilatation of the lateral and third ventricles. The most common cause of obstructive (noncommunicating) hydrocephalus COMMUNICATING HYDROCEPHALUS--interference of absorption of CSF due to either 1) occlusion of the subarachnoid cisterns around the brainstem or 2) to obliteration of the subarachnoid spaces over the convexities of the brain. Therefore, the entire ventricular system is uniformly distended. DANDY-WALKER MALFORMATION--See Dandy-Walker malformation abstraction table OTHER ANATOMIC LESIONS--includes hydrocephalus due to encephaloceles, agenesis of the corpus callosum, arachnoid and interhemispheric cysts, and occlusion of the foramen of Monro (see exclusions below)
ICD-9-CM CODES	742.3
INCLUSIONS	primarily includes cases whose etiology is aqueductal stenosis or with unknown etiology
EXCLUSIONS	--most cases with communicating hydrocephalus (e.g., due to intraventricular hemorrhage) --hydrocephalus due to other anatomic lesions unless the primary lesion is an inclusion diagnosis such as holoprosencephaly (if the primary lesion is an inclusion diagnosis, the case is eligible; however, hydrocephalus as a defect should be excluded by checking the "Eligible Defect but does not meet NBDPS Criteria" box.) --cases associated with bone dysplasias --lesions described as hydrocephalus ex vacuo, colpocephaly, ventriculomegaly, hydranencephaly --cases not documented by postnatal brain imaging (head ultrasound, CT or MRI scan), neurosurgical operative report or autopsy
ADDITIONAL BIRTH DEFECTS	central nervous system defects such as Chiari II malformation and neural tube defects (spina bifida cystica, encephalocele); Klippel-Feil anomaly
INFORMATION SOURCES-MED. RECORDS	-- prenatal ultrasound, clinicians' or nurses' notes (signs can include sunsetting eyes, tense fontanelle) --diagnosis confirmed by postnatal head ultrasound, CT or MRI scan of the brain --surgical notes, autopsy/pathology reports, if done GOLD STANDARD(S): CT
DEFECT-SPECIFIC INFORMATION TO ABSTRACT	1) head circumference and percentiles 2) viral infections (maternal history of intrauterine infections); viral titers and cultures, if done 3) verbatim description of brain imaging studies
ADD. COMMENTS	HYDRO: water, fluid; CEPHALY: head Hydrocephalus has heterogenous etiologies with genetic and nongenetic causes including infectious agents, intracranial hemorrhage, or tumors. NOTE: If you have a question or the diagnosis seems unclear, record all information and clinical reviewer will make the final decision.

Hypospadias, second- or third-degree

BIRTH DEFECT & DEFINITION	SECOND- OR THIRD-DEGREE HYPOSPADIAS--displacement of the urethral meatus ventrally and proximally from the tip of the glans penis. The degree is classified according to the position of the meatus on the penile shaft. In about 35-50% of the cases, there is associated chordee which is the downward curvature of the glans penis with resultant penile deformity.
TYPES & DEFINITIONS	FIRST-DEGREE--urethral meatus on the glans or corona (glandular, coronal) SECOND-DEGREE--urethral meatus on the shaft (penile) THIRD-DEGREE--urethral meatus on the scrotum or perineum (scrotal, perineal)
ICD-9-CM CODES	752.61
INCLUSIONS	standard
EXCLUSIONS	first-degree hypospadias; hypospadias, NOS; chordee alone; epispadias; ambiguous genitalia without further description (see comments below)
ADDITIONAL BIRTH DEFECTS	chordee (can be cutaneous or fibrous), genitourinary tract abnormalities
INFORMATION SOURCES-MED. RECORDS	clinicians' and nurses' notes, urology consult, possibly endocrinology and genetics consults, operative/path reports, autopsy, possibly radiographic studies such as voiding cystourethrogram or retrograde urethrocystography
DEFECT-SPECIFIC INFORMATION TO ABSTRACT	degree of hypospadias; description of location of urethral meatus
ADD. COMMENTS	Third-degree hypospadias may initially be described as ambiguous genitalia; careful chart review may be necessary to obtain final diagnosis.

Intestinal atresia/stenosis

BIRTH DEFECT & DEFINITION	INTESTINAL ATRESIA/STENOSIS--complete or partial occlusion of the lumen of a segment of small or large intestine. Single or multiple areas of occlusion can occur.
TYPES & DEFINITIONS	DUODENAL ATRESIA/STENOSIS--affected area is the duodenum JEJUNAL ATRESIA/STENOSIS--affected area is the jejunum ILEAL ATRESIA/STENOSIS--affected area is the ileum LARGE INTESTINAL ATRESIA/STENOSIS--affected area is the large intestine RECTAL ATRESIA/STENOSIS--affected area is the rectum ANAL ATRESIA/IMPERFORATE ANUS--affected area is the anus NOTE: these defects can occur with or without a fistula
ICD-9-CM CODES	ATRESIA/STENOSIS OF SMALL INTESTINE--751.1 ATRESIA/STENOSIS OF LARGE INTESTINE, RECTUM, AND ANAL CANAL--751.2
INCLUSIONS	standard
EXCLUSIONS	--anal stenosis, anteriorly displaced anus --intestinal atresia/stenosis in an infant with cystic fibrosis --duodenal atresia associated with duodenal web or with annular pancreas
ADDITIONAL BIRTH DEFECTS	annular pancreas and rotational abnormalities, (e.g., volvulus, malrotation) "apple peel atresia"--distinct type of jejunal atresia; commonly has associated anomalies VATER/VACTERL association--Vertebral defects, Anal atresia, Cardiac defects, T-E fistula, Renal defects, Limb anomalies Important to note if single umbilical artery is present
INFORMATION SOURCES-MED. RECORDS	prenatal ultrasound, postnatal x-ray ("double bubble" sign characteristic of duodenal atresia), barium enema, clinicians' or nurses' notes, surgery consult, operative/path reports, autopsy GOLD STANDARD(S): barium enema, operative/path reports, autopsy
DEFECT-SPECIFIC INFORMATION TO ABSTRACT	presence of abdominal wall defect--omphalocele or gastroschisis
ADD. COMMENTS	--If contiguous regions of the small intestine are involved, a compound descriptor may be used, e.g., jejunoileal atresia. Codes for both affected areas should be included, but the descriptor should indicate whether these are contiguous or non-contiguous regions. One-third of all infants with duodenal atresia/stenosis have Down syndrome.

Limb deficiency, intercalary

BIRTH DEFECT & DEFINITION	INTERCALARY LIMB DEFICIENCY--complete or partial absence of proximal or middle segment(s) of a limb with all or part of the distal segment present OTHER NAMES: phocomelia DEFECT GROUP: Limb deficiency or reduction
TYPES & DEFINITIONS	PHOCOMELIA--used to designate any intercalary deficiency (complete, proximal, and distal have been used to designate deficiencies of the humeral and radioulnar, humeral, and radioulnar components, respectively) HUMERAL/FEMORAL DEFICIENCY--complete or partial absence of the humerus or femur with presence of some of the distal limb elements RADIOULNAR/TIBIOFIBULAR DEFICIENCY--complete or partial absence of the radius/ulna or tibia/fibula with presence of some of the distal limb elements
ICD-9-CM CODES	755.23-755.25, 755.33-755.35
INCLUSIONS	standard
EXCLUSIONS	unspecified limb deficiency, generalized limb shortening including chondrodysplasias, nail hypoplasia
ADDITIONAL BIRTH DEFECTS	positional abnormalities of distal structures may be present FEMUR-FIBULA-ULNA SYNDROME (FFU) or PROXIMAL FEMORAL FOCAL DEFICIENCY (PFFD) is characterized by defects of the femur, fibula, ulna
INFORMATION SOURCES-MED. RECORDS	prenatal ultrasound, clinicians' (especially orthopedics, genetics) and nurses' notes, x-ray, surgical or autopsy notes GOLD STANDARD(S): x-ray of affected limbs (but keep in mind that not all bones in hands/feet are ossified at this age, therefore, "not seen" does not necessarily mean "not present")
DEFECT-SPECIFIC INFORMATION TO ABSTRACT	1) laterality 2) verbatim x-ray report
ADD. COMMENTS	The term "phocomelia" is misused. The best description of the defect includes a complete description of all absent and present structures. Thalidomide is classically associated with phocomelia, however, radial ray abnormalities may be just as common in this

Limb deficiency, longitudinal

BIRTH DEFECT & DEFINITION	LONGITUDINAL LIMB DEFICIENCY--partial absence of a limb extending parallel with the long axis of the limb and involving the preaxial, postaxial, or central components DEFECT GROUP: Limb deficiency or reduction
TYPES & DEFINITIONS	RADIAL OR TIBIAL APLASIA/HYPOPLASIA--complete or partial absence of the portion of the forearm, hand, leg or foot on the radial (thumb) or tibial (great toe/hallux) side of the limb OTHER NAMES: preaxial limb deficiency or reduction, tibial hemimelia, radial agenesis ULNAR OR FIBULAR APLASIA/HYPOPLASIA--complete or partial absence of the portion of the forearm, hand, leg, or foot on the ulnar or fibular side of the limb OTHER NAMES: postaxial limb deficiency or reduction, ulnar hemimelia, ulnar agenesis SPLIT-HAND/SPLIT-FOOT MALFORMATION (SHSF)--complete or partial absence of one or more of the central rays of the hand or foot (fingers/toes and metacarpals/metatarsals 2-4, or monodactyly) OTHER NAMES: ectrodactyly, ectromelia, lobster claw hand
ICD-9-CM CODES	755.26-755.27, 755.36-755.37, no specific code for SHSF MALFORMATION (uses 755.4)
INCLUSIONS	Includes isolated missing thumb: use "thumb only" codes. When an absent thumb is accompanied by a radial deficiency, a radial aplasia/hypoplasia code alone can be used for both the thumb and radial deficiencies.
EXCLUSIONS	unspecified limb deficiency, generalized limb shortening including chondrodysplasias, nail hypoplasia
ADDITIONAL BIRTH DEFECTS	LIMB DEFECTS--tibial aplasia may also be associated with preaxial polydactyly; SHSF is frequently associated with tibia aplasia; clubfoot or other joint contractures are common with limb deficiencies; bowing or other distortion of the ulna without deficiency may be present with severe radial hypoplasia or aplasia ANY DEFECT--VACTERL association has Vertebral anomalies, Anal atresia, Cardiac defects, T-E fistula, Renal anomalies, Limb anomalies
INFORMATION SOURCES-MED. RECORDS	prenatal ultrasound, clinicians' (especially orthopedics, genetics) and nurses' notes, x-ray, surgical or autopsy notes GOLD STANDARD(S): x-ray of affected limbs (but keep in mind that not all bones in hands/feet are ossified at this age, therefore, "not seen" does not necessarily mean "not present")
DEFECT-SPECIFIC INFORMATION TO ABSTRACT	1) laterality 2) verbatim x-ray reports
ADD. COMMENTS	The best description of the defect includes a complete description of all absent and pertinent present structures. The term "ectrodactyly" is also sometimes used to describe a transverse terminal defect.

Limb deficiency, transverse

BIRTH DEFECT & DEFINITION	TRANSVERSE LIMB DEFICIENCY--complete or partial absence of distal structures of a limb in a transverse plane at the point where the deficiency begins with proximal structures essentially intact. (Proximal--nearest the trunk or point of origin. Distal--away from the trunk or point of origin.) OTHER NAMES: congenital amputation DEFECT GROUP: Limb deficiency or reduction
TYPES & DEFINITIONS	AMELIA--complete absence of a limb HEMI- OR MEROMELIA--partial absence of a limb (rather nonspecific and can be used for longitudinal defects also) TRANSVERSE TERMINAL DEFICIENCY--absence of distal structures with proximal structures essentially intact (used for deficiencies below the elbow) APHALANGIA--absence of phalanges; ADACTYLY--absence of digits; OLIGODACTYLY--fewer than 5 digits; ACHEIRIA--absence of a hand
ICD-9-CM CODES	755.21, 755.29, 755.31, 755.39
INCLUSIONS	isolated missing digits except isolated missing thumb
EXCLUSIONS	unspecified limb deficiency, generalized limb shortening including chondrodysplasias, nail hypoplasia
ADDITIONAL BIRTH DEFECTS	POLAND ANOMALY (transverse terminal defect with ipsilateral pectoral muscle deficiency) MOEBIUS ANOMALY, OROMANDIBULAR-LIMB HYPOGENESIS SPECTRUM (small mouth, micrognathia, hypoglossia, oligodactyly, hypoplastic limbs, 6th and 7th cranial nerve palsy)
INFORMATION SOURCES-MED. RECORDS	prenatal ultrasound, clinicians' (especially orthopedics, genetics) and nurses' notes, x-ray, surgical or autopsy notes GOLD STANDARD(S): x-ray of affected limbs (but keep in mind that not all bones in hands/feet are ossified at this age, therefore, "not seen" does not necessarily mean "not present")
DEFECT-SPECIFIC INFORMATION TO ABSTRACT	1) amniotic band sequence 2) verbatim x-ray report 3) laterality
ADD. COMMENTS	Rudimentary or nubbin fingers may be found at the distal end of a transverse deficiency. The best description of a defect includes a complete description of all absent and pertinent present structures.

Limb deficiency, NEC

BIRTH DEFECT & DEFINITION	NEC--refers to any limb deficiency not elsewhere classifiable. Please refer to other limb abstraction instruction forms for more information
TYPES & DEFINITIONS	Please refer to other limb abstraction instruction forms for more information
ICD-9-CM CODES	Please refer to other limb abstraction instruction forms for more information
INCLUSIONS	
EXCLUSIONS	
ADDITIONAL BIRTH DEFECTS	Please refer to other limb abstraction instruction forms for more information
INFORMATION SOURCES-MED. RECORDS	Please refer to other limb abstraction instruction forms for more information
DEFECT-SPECIFIC INFORMATION TO ABSTRACT	1) laterality 2) verbatim x-ray reports
ADD. COMMENTS	The best description of the defect includes a complete description of all absent and pertinent present structures.

Obstructive heart defects

BIRTH DEFECT & DEFINITION	OBSTRUCTIVE HEART DEFECTS--congenital heart defects in which there is obstruction to the flow of blood from either the left or right side of the heart or the great vessels.
TYPES & DEFINITIONS	LEFT-SIDED OBSTRUCTIVE ANOMALIES: AORTIC STENOSIS, VALVAR (ASV)--Obstruction or narrowing of the aortic valve. NOTE: Aortic stenosis can also occur above the valve (supra-valvar) or below the valve (sub-valvar). Both of these defects are excluded from the study. COARCTATION OF THE AORTA (COA)--Narrowing of the descending aorta, distal to the left subclavian, usually in the region where the ductus arteriosus inserts. NOTE: Precise designation of COA location as "pre-", "juxta-", or "postductal" is unnecessary. Most are juxtaductal. Most COAs involve a discrete ledge but some are long segment. For this project, include both, although they are probably mechanistically different. INTERRUPTED AORTIC ARCH, TYPE A (IAA, TYPE A)--Interruption of the descending aorta, distal to the left subclavian artery. Like a COA but so severe that there is interruption. NOTE: IAA, type B is classified as a conotruncal heart defect and is more common than type A. If type A or B are not specified, please note location of the lesion (proximal or distal to the subclavian artery) for the clinical reviewer. HYPOPLASTIC LEFT HEART SYNDROME (HLHS)--Extreme smallness of the left heart structures and aorta. Classically included mitral atresia/severe hypoplasia, aortic atresia/severe hypoplasia, aorta hypoplasia, and coarctation. For the purposes of case-definition for the study (classic HLHS), the first two anomalies must be present. NOTE: Most cases fulfill the classic definition but sometimes HLHS is used broadly, e.g. DORV or AVC with very small left ventricle. Record these anomalies in their specific categories not as HLHS. For the purpose of the study, with classic HLHS coarctation should be coded separately, but the mitral and aortic atresia/hypoplasia descriptions can be included under the HLHS code. RIGHT-SIDED OBSTRUCTIVE ANOMALIES: PULMONIC STENOSIS, VALVAR (PSV)--Obstruction or narrowing of the pulmonic valve. NOTE: Pulmonic stenosis can also occur above the valve (supra-valvar) or below the valve (sub-valvar). The main or branch pulmonary arteries can also be stenotic (PAS). Only valvar stenosis is included in the study. Cases in which "pulmonic stenosis" is specified on echocardiogram, but without additional information regarding site of stenosis, should be assumed to be valvar pulmonic stenosis for the purposes of this study. PULMONARY ATRESIA/INTACT VENTRICULAR SEPTUM (PA/IVS)--Absent connection between the right ventricle and the pulmonary artery. Pulmonary valve may be present, but not patent (atresia), or may not have formed at all (agenesis). NOTE: This defect differs markedly from pulmonary atresia with VSD, which is usually more like Tetralogy of Fallot (see conotruncal heart defects). However, if PA/VSD are both present and anatomic details of the VSD/aorta are not described in the echocardiogram report, e.g., "membranous/malalignment-type," or if the VSD is described as "muscular," the nonspecific Pulmonary Atresia with VSD (not TOF variant 747.310) code should be used. TRICUSPID ATRESIA (TATR, TAT, TRA)--Absent connection between the right atrium and the right ventricle. NOTE: Tricuspid valve may be present but closed (atresia), or completely absent (agenesis). Usually accompanied by other CHDs.
ICD-9-CM CODES	ASV--746.3; COA--747.10; IAA, TYPE A--747.11; HLHS--746.7 PS--746.02; PA/IVS--746.00; TAtr--746.1
INCLUSIONS	standard
EXCLUSIONS	coarctation of the aorta cases prenatally diagnosed that do not have a postnatal examination to confirm the diagnosis
ADDITIONAL BIRTH DEFECTS	Just about any defect can be found with these CHDs. HLHS is often seen as an isolated defect.
INFORMATION SOURCES-MED. RECORDS	clinicians' or nurses' notes, pediatric cardiology consultation, pathology reports, cardiology diagnostic reports. GOLD STANDARD(S): echo (less reliable is prenatal ultrasound/echo), cath, surgery, autopsy
DEFECT-SPECIFIC INFORMATION TO ABSTRACT	standard
ADD. COMMENTS	Along with the septal defects, these obstructive lesions are classified as Clark's Class III. Although the obstructive lesions of the left heart (ASV, COA, HLHS) are more heterogeneous, they do "hang together" strongly. Turner syndrome (45,X or variants) is associated with left-heart CHDs. Noonan syndrome is associated with PSV.

Omphalocele

BIRTH DEFECT & DEFINITION	OMPHALOCELE--a defect in the anterior abdominal wall, accompanied by herniation of some abdominal organs through a widened umbilical ring, into the umbilical stalk. The defect is covered by a thin, nearly transparent sac composed of the amnion and lined by peritoneum. The umbilical cord inserts into the sac. Sac is RARELY ruptured. OTHER NAMES: exomphalos, amniocele
TYPES & DEFINITIONS	PENTALOGY OF CANTRELL--1) supraumbilical midline abdominal wall defect, 2) defect of lower sternum, 3) deficiency of anterior diaphragm, 4) defect in diaphragmatic pericardium 5) congenital heart defects OEIS COMPLEX--Omphalocele-Exstrophy-Imperforate anus-Spina bifida
ICD-9-CM CODES	756.79
INCLUSIONS	all cases including those prenatally diagnosed if high resolution u/s was done and umbilicus was visualized
EXCLUSIONS	umbilical hernia (rarely present at birth); must be distinguished from gastroschisis
ADDITIONAL BIRTH DEFECTS	lung hypoplasia
INFORMATION SOURCES-MED. RECORDS	prenatal ultrasound, clinicians' or nurses' notes, autopsy, surgery notes GOLD STANDARD(S): surgery notes or autopsy NOTE: maternal serum and/or amniotic fluid alpha fetoprotein may be elevated
DEFECT-SPECIFIC INFORMATION TO ABSTRACT	1) size (diameter) and location of abdominal wall defect 2) # of umbilical vessels 3) which organs are herniated into the umbilicus 4) describe sac (intact?, bowel matted?)
ADD. COMMENTS	--Umbilical hernia has complete skin covering, no thin transparent sac. --It is essential to differentiate omphalocele from gastroschisis, in which the protruding abdominal contents were floating in amniotic fluid, and the bowel is often matted and adhered together by a fibrous mat. In gastroschisis, the extruded abdominal contents may be matted and covered by a thick fibrous material, but this membrane does not resemble skin. Gastroschisis occurs to the side of the umbilicus.

Renal agenesis/hypoplasia

BIRTH DEFECT & DEFINITION	RENAL AGENESIS--complete absence of the kidney RENAL HYPOPLASIA--incomplete development of the kidney OTHER NAMES: renal aplasia, renal dysgenesis, Potter syndrome
TYPES & DEFINITIONS	N/A
ICD-9-CM CODES	753.0
INCLUSIONS	--only bilateral cases --prenatally-diagnosed only cases if there is no visualization of the kidneys AND oligohydramnios --bilateral hypoplasia--which is severe enough to cause oligohydramnios
EXCLUSIONS	--renal dysplasia or cystic dysplasia --unilateral renal agenesis with contralateral multicystic dysplasia --"small kidneys"
ADDITIONAL BIRTH DEFECTS	OLIGOHYDRAMNIOS SEQUENCE--consequence of decreased amniotic fluid and consists of minor facial dysmorphia (flat face, small chin, large ears), pulmonary hypoplasia, joint contractures OTHER UROGENITAL ANOMALIES--absent or abnormal ureters, bladder, vagina, uterus, seminal vesicles, or vas deferens; undescended testes, MURC association (MÜllerian duct anomalies, Renal agenesis, Cervical vertebral anomalies) OTHER--single umbilical artery, spina bifida, VACTERL association (Vertebral anomalies, Anal atresia, Cardiac defects, T-E fistula, Renal anomalies, Limb anomalies)
INFORMATION SOURCES-MED. RECORDS	pre- or postnatal ultrasound, autopsy, occasionally surgery, CT scan or MR studies GOLD STANDARD(S): autopsy NOTE: renal scans or intravenous pyelograms (IVP) may be unreliable since uptake can be minimal or none with a severely dysplastic kidney
DEFECT-SPECIFIC INFORMATION TO ABSTRACT	1) prenatal diagnosis--may show early hydronephrosis which could destroy renal tissue and be more properly classified as renal dysplasia 2) verbatim descriptions in imaging studies, autopsy, weight of kidneys (if available)
ADD. COMMENTS	It is often difficult to separate out the renal dysplasias. Abstract if in doubt and leave the decision for the clinical reviewer.

Sacral agenesis

BIRTH DEFECT & DEFINITION	SACRAL AGENESIS--agenesis of terminal segments of vertebral column, ranging from absence of lower coccygeal segments to complete aplasia of vertebrae below 12th thoracic segment. OTHER NAMES: sacral regression, caudal regression
TYPES & DEFINITIONS	Types I-IV are described and are based on the extent of the defect.
ICD-9-CM CODES	756.13
INCLUSIONS	standard
EXCLUSIONS	sirenomelia
ADDITIONAL BIRTH DEFECTS	vertebral defects including cervical vertebrae; pelvic defects; lower extremity defects such as hip and knee contractures and clubfoot; GI defects; GU defects; defects consistent with the VACTERL association (Vertebral anomalies, Anal atresia, Cardiac defects, T-E fistula, Renal defects, and Limb defects)
INFORMATION SOURCES-MED. RECORDS	prenatal ultrasound, clinicians' and nurses' notes, X-rays, ultrasound, CT, MRI, surgical notes, autopsy GOLD STANDARD(S): X-rays, physical exam, particularly from a geneticist
DEFECT-SPECIFIC INFORMATION TO ABSTRACT	Is the mother diabetic? (Note which class, if stated in chart)
ADD. COMMENTS	none

Septal heart defects

BIRTH DEFECT & DEFINITION	SEPTAL HEART DEFECTS--defects of the septum dividing the atria or ventricles
TYPES & DEFINITIONS	MUSCULAR (TYPE IV) VENTRICULAR SEPTAL DEFECT (VSDMUS)--Hole (defect) in the muscular ventricular septum which comprises the bulk of the septum. Can be single or multiple ("Swiss cheese septum"), tiny or large. PERIMEMBRANOUS (TYPE II, MEMBRANOUS) VENTRICULAR SEPTAL DEFECT (VSDMEM)--Hole (defect) in the membranous portion of the ventricular septum. "Aneurysm of ventricular septum" is a closing VSDmem. INFLOW-TYPE (SUBTRICUSPID, CANAL-TYPE) VENTRICULAR SEPTAL DEFECT (VSDAVC)--Discussed as part of the atrioventricular (AV) canal or endocardial cushion defect spectrum (see atrioventricular septal defects abstraction sheet). ATRIAL SEPTAL DEFECT, SECUNDUM TYPE (ASD2)--Hole (defect) usually in the midportion of the atrial septum. Must be distinguished from ASD primum, which is lower in the septum, and from patent foramen ovale, which is a physiologic opening in the septum but not a defect. VENTRICULAR SEPTAL DEFECT, NOT OTHERWISE SPECIFIED (VSD, NOS)--Ventricular septal defects in which the record does not specify a particular location in the septum. These VSDs are usually VSDmem. ATRIAL SEPTAL DEFECT, NOT OTHERWISE SPECIFIED (ASD, NOS)--Atrial septal defects in which the record does not specify a particular location in the septum. Vast majority of ASD, NOS are ASD2 in contrast to the primum or rare sinoseptal types. Some small ASD2 cannot be distinguished from large patent foramen ovale (PFO).
ICD-9-CM CODES	VSDMUS; VSDMEM; VSD, NOS--745.4; VSDAVC--745.69; ASD2--745.5
INCLUSIONS	ASD, NOS will be included as ASD2
EXCLUSIONS	--muscular VSDs in infants < 37 weeks gestation --diagnosis of ASD versus PFO (see additional comments) --Because of the large number of VSDs, the NBDPS has instituted a moratorium on the collection of muscular VSDs and VSDs, NOS (cases in which these types of VSDs are the sole eligible defects), beginning with birth dates on or after 10/1/98 for Centers beginning the study on 10/1/97, and on or after 1/1/99 for Centers beginning the study on 1/1/98. In cases in which the diagnosis states "VSD, NOS", efforts should continue to be made to determine the specific type of defect, since other types of VSDs (membranous, malalignment, etc.) will still be included in the NBDPS, as before the institution of this moratorium. In addition, cases with VSDs (NOS and muscular) which also have other eligible defects will continue to be collected, as before.
ADDITIONAL BIRTH DEFECTS	Just about any defect can be found with these CHDs.
INFORMATION SOURCES-MED. RECORDS	clinicians' or nurses' notes, pediatric cardiology consultation, pathology reports, cardiology diagnostic reports. GOLD STANDARD(S): echo (less reliable is prenatal ultrasound/echo), cath, surgery, autopsy
DEFECT-SPECIFIC INFORMATION TO ABSTRACT	standard
ADD. COMMENTS	--Cases with the diagnosis ASD vs. PFO should be collected for clinician review; if the diagnosis cannot be established, these cases will be excluded from the study. --Roger's disease and Eisenmenger's syndrome are archaic terms and should not be used to describe a VSD. --Most defects in this group fall in Class II of Clark's classification (attributed to altered embryonic intracardiac blood flow). Muscular VSDs are in Class III (putatively attributed to abnormal cell death).

Single ventricle

BIRTH DEFECT & DEFINITION	SINGLE VENTRICLE--essentially only one functioning ventricle. This is always a complex heart and most commonly is a double inlet left ventricle. OTHER NAMES: common ventricle, cor triloculare biatriatum (archaic) NOTE: Usually the 'single ventricle' coexists with other heart malformation so that overall the heart malformations are very complex. Coexisting heart defects include malposed great arteries, rudimentary outlet chambers (the tiny second ventricle), often PS, COA.
TYPES & DEFINITIONS	It is important to identify the morphology of the functional ventricle (left, right, or indeterminate), and the connection between the atria and the ventricles (both valves connected to the same one ventricle vs. one AV valve per ventricle). A second ventricle is often present, albeit rudimentary and non-functional. It is customary to split the 'single ventricle' category in subtypes: DOUBLE INLET LEFT VENTRICLE--the functional ventricle is morphologically 'left' and both AV valves open into it. This is the most common type. DOUBLE INLET RIGHT VENTRICLE--same as above but the functional ventricle is morphologically 'right'. This is rare. DOUBLE INLET VENTRICLE OF INDETERMINATE MORPHOLOGY-- when ventricular morphology is indeterminate. This type needs to be differentiated from those which are unspecified or NOS.
ICD-9-CM CODES	745.3
INCLUSIONS	standard
EXCLUSIONS	standard
ADDITIONAL BIRTH DEFECTS	Just about any defect can be found with these CHDs.
INFORMATION SOURCES-MED. RECORDS	clinicians' or nurses' notes, pediatric cardiology consultation, pathology reports, cardiology diagnostic reports. GOLD STANDARD(S): echo (less reliable is prenatal ultrasound/echo), cath, surgery, autopsy
DEFECT-SPECIFIC INFORMATION TO ABSTRACT	standard
ADD. COMMENTS	Other situations, such as hypoplastic left heart syndrome and tricuspid atresia, are superficially similar to single ventricle in that one ventricle is extremely small and nonfunctional. These forms, however, are due to atresia of one of the AV valves and should be coded using the hypoplastic left heart and tricuspid atresia codes, respectively. This defect does not have a specific Clark classification; it is a very heterogeneous condition.

NBDPS Abstractor's Instructions

Spina bifida

BIRTH DEFECT & DEFINITION	SPINA BIFIDA--herniation of the meninges and/or spinal cord tissue through a bony defect of spine closure OTHER NAMES: spina bifida cystica, spina bifida aperta, myeloschisis, myelodysplasia, etc., etc. DEFECT GROUP: neural tube defects (NTDs)
TYPES & DEFINITIONS	MENINGOMYELOCELE/MYELOMENIGOCELE--90% of lesions, herniation of meninges and spinal cord tissue MENINGOCELE--herniation of meninges without spinal cord tissue RACHISCHISIS--spine defect without meninges covering the neural tissue LIPOMENINGOMYELOCELE/LIPOMENINGOCELE--lipomatous (fatty) tissue associated with a bony defect of the spine and herniation of meninges or spinal cord tissue, usually located in the lumbar region MYELOCYSTOCELE--cystic lesion of the spinal cord central canal and herniation through a spinal defect OPEN LESION--neural tissue open to environment or covered by membrane only (90% of lesions) CLOSED LESION--neural tissue covered by normal skin LEVEL OF LESION--highest and lowest vertebrae--cervical (C), thoracic (T), lumbar (L), sacral (S)
ICD-9-CM CODES	SPINA BIFIDA--741.0-741.9
INCLUSIONS	all cases including those cases prenatally diagnosed that do not have a postnatal examination to confirm the defect
EXCLUSIONS	spina bifida occulta, primary tethered cord, syringomyelia (hydromyelia), diastematomyelia, diplomyelia
ADDITIONAL BIRTH DEFECTS	hydrocephalus, Arnold-Chiari II malformation, agenesis of the corpus callosum, hydronephrosis, renal agenesis, rib anomalies, clubfoot, developmental dysplasia of the hip, scoliosis, tethered cord (usually acquired)
INFORMATION SOURCES-MED. RECORDS	prenatal ultrasound, clinicians' or nurses' notes, surgery notes, autopsy, x-ray, CT or MRI scans GOLD STANDARD(S): TYPE--can rarely be determined by inspection (exception: rachischisis), surgical or autopsy report, MR or CT scan; LEVEL OF LESION--surgical or autopsy report, MR or CT scan, spine x-ray; OPEN/CLOSED STATUS--clinicians' or nurses' notes, surgery notes, autopsy NOTE: 1) maternal serum and amniotic fluid alpha-fetoprotein and acetylcholinesterase will be elevated in open lesions since neural tissue is in contact with the amniotic fluid 2) sterile saline gauze is often applied to open lesions in the delivery room even if not leaking CSF
DEFECT-SPECIFIC INFORMATION TO ABSTRACT	1) Was the lesion leaking or was special nursing care required? (indicates open lesion) 2) Elements of a complete description: type, location (verbatim level), open/closed status, other CNS anomalies. Example--Open meningomyelocele from T2-T8 with an Arnold-Chiari II malformation and hydrocephalus
ADD. COMMENTS	MENINGO: membrane; MYELO: spinal cord; RACHIS: spine; SCHISIS: fissure; LIPO: fat; APERTA: open; CELE: hernia NOTE: Names for types of spina bifida are numerous; only most common are listed above